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Tetraploid human sperm produced in the absence of meiotic divisions I and II were detected by multicolor FISH for chromosomes X, Y, and 21 and by phase contrast microscopy

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During normal spermatogenesis, primary spermatocytes give rise to sperm after completing two meiotic divisions. We present FISH cytogenetic and morphological evidence that primary spermatocytes may differentiate into seminal sperm without undergoing either meiotic divisions. Phase contrast microscopy and multi-color FISH for chromosomes X, Y, and 21 was used to identify flagellated cells carrying the fluorescence genotype of a primary spermatocyte, i.e., X-X-Y-Y-21-21-21-21. Such tetraploid cells typically have sperm-like nuclear and fluorescence-domain morphologies. Tetraploid sperm were generally larger than normal sperm and had multiple tails. These sperm were consistently found among 8 semen samples provided by five patients, ~40 to ~100 days after they were exposed to ultra-high doses of diazapam during suicide attempts (1.4 ± 1.2 per 10,000 cells; >80,000 cells). However, none of these cells were detected among >50,000 cells from five healthy men. Previously, we showed that these patients also produced increased frequencies of hyperhaploid and diploid sperm (Baumgartner et al, 1996). Our findings suggest that (a) the completion of both meiosis I and II is not a requirement for the formation of a human sperm and (b) human spermiogenesis can package a large nucleus with four times the haploid set of chromosomes into a sperm.

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